ALDH3A2 gene

aldehyde dehydrogenase 3 family member A2

Normal Function

The *ALDH3A2* gene is a member of the aldehyde dehydrogenase (ALDH) gene family. Genes in this family provide instructions for producing enzymes that alter molecules called aldehydes. The *ALDH3A2* gene provides instructions for making an enzyme called fatty aldehyde dehydrogenase (FALDH). This enzyme is involved in the breakdown of fats, specifically the breakdown of molecules called fatty aldehydes to fatty acids. This conversion of molecules is part of a multistep process called fatty acid oxidation in which fats are broken down and converted into energy.

The FALDH enzyme is found in most tissues, but its activity (expression) is highest in the liver. Within cells, the FALDH enzyme is located in the endoplasmic reticulum, a structure involved in protein processing and transport.

Health Conditions Related to Genetic Changes

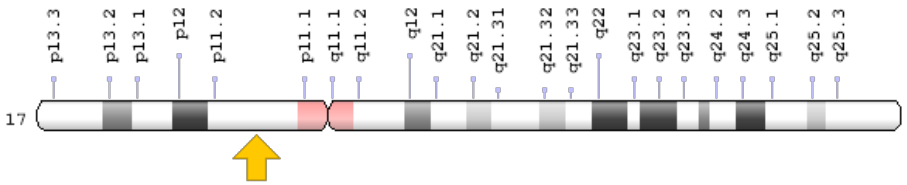
Sjögren-Larsson syndrome

At least 80 mutations in the *ALDH3A2* gene have been found to cause Sjögren-Larsson syndrome, a condition characterized by dry, scaly skin (ichthyosis); neurological abnormalities; and eye problems. Many of these mutations change single protein building blocks (amino acids) in the FALDH enzyme. The gene mutations that cause Sjögren-Larsson syndrome lead to the production of a FALDH enzyme that is unable to break down fatty aldehyde molecules. As a result, fats that are not broken down can build up in cells. In all affected tissues, excess fat accumulation interferes with the normal formation of protective membranes or materials that are necessary for the body to function normally. These abnormalities underlie the characteristic signs and symptoms of Sjögren-Larsson syndrome.

Chromosomal Location

Cytogenetic Location: 17p11.2, which is the short (p) arm of chromosome 17 at position 11.2

Molecular Location: base pairs 19,648,136 to 19,677,596 on chromosome 17 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- AL3A2_HUMAN
- aldehyde dehydrogenase 3 family, member A2
- aldehyde dehydrogenase 10
- aldehyde dehydrogenase family 3 member A2
- ALDH10
- FALDH
- fatty aldehyde dehydrogenase
- microsomal aldehyde dehydrogenase

Additional Information & Resources

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28ALDH3A2%5BTIAB%5D%29+OR+%28FALDH%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2520+days%22%5Bdp%5D

OMIM

- ALDEHYDE DEHYDROGENASE, FAMILY 3, SUBFAMILY A, MEMBER 2
  http://omim.org/entry/609523

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_ALDH3A2.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=ALDH3A2%5Bgene%5D
- HGNC Gene Family: Aldehyde dehydrogenases
  http://www.genenames.org/cgi-bin/genefamilies/set/398
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=403
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/224
- UniProt
  http://www.uniprot.org/uniprot/P51648

Sources for This Summary

- OMIM: ALDEHYDE DEHYDROGENASE, FAMILY 3, SUBFAMILY A, MEMBER 2
  http://omim.org/entry/609523
- Ashibe B, Motojima K. Fatty aldehyde dehydrogenase is up-regulated by polyunsaturated fatty acid via peroxisome proliferator-activated receptor alpha and suppresses polyunsaturated fatty acid-induced endoplasmic reticulum stress. FEBS J. 2009 Dec;276(23):6956-70. doi: 10.1111/j.1742-4658.2009.07404.x. Epub 2009 Oct 27.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19860831
- Rizzo WB, S'Aulis D, Jennings MA, Crumrine DA, Williams ML, Elias PM. Ichthyosis in Sjögren-Larsson syndrome reflects defective barrier function due to abnormal lamellar body structure and secretion. Arch Dermatol Res. 2010 Aug;302(6):443-51. doi: 10.1007/s00403-009-1022-y. Epub 2010 Jan 5.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20049467
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2892059/
- Rizzo WB. Sjögren-Larsson syndrome: molecular genetics and biochemical pathogenesis of fatty aldehyde dehydrogenase deficiency. Mol Genet Metab. 2007 Jan;90(1):1-9. Epub 2006 Sep 22. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16996289
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1933507/

Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/gene/ALDH3A2

Reviewed: October 2011
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services